

Emergency Metabolic Protocol

RE:
DOB:

Please see this patient immediately.

_____ has Maple Syrup Urine Disease (MSUD), an inherited metabolic disorder of branched chain amino acid metabolism characterized by the buildup of amino and organic acids. The amino acids accumulating in excess in this disease can cause brain edema with alteration of sensorium progressing to coma, herniation of cerebellar tonsils and death. The disease is controlled by a special diet low in protein and branched chain amino acids. The disorder worsens in case of fasting or fever/infection, when the body needs to use endogenous proteins.

In case of acute deterioration, the following laboratory studies should be obtained:

Basic Metabolic Panel
CBC with differential
Urine analysis
Urine organic acids
Plasma amino acids

The patient should receive IV fluids consisting of:

D10 Normal Saline 20 mEq/L KCl at maintenance rate
Intralipids 20% at 4g/kg/day (20ml/kg/day)
These will provide calories to prevent catabolism and decrease the protein load in his system

If admitted, consider placing a PICC line for IV fluid administration to prevent multiple IV placements

Glucose should be monitored every hour for 4 hours after starting the infusion. If glucose is >160, insulin at 0.1 U/kg should be given as bolus. Insulin is the single most effective way of decreasing leucine levels. Insulin can be repeated at the same dose every hour if glucose remains elevated and should be stopped when glucose drops below 80 mg/dL.

BMP should be monitored Q4h until the child is responsive and alert.

Sodium should be maintained at or above 135 mM to reduce the risk for brain edema. Consider using IV mannitol (0.5 g/kg per dose) in case of suspected brain edema (even before the IV fluids). If sodium remains <135 mM, give NaCl 3% 1 ml per kilogram.

Enteral feedings with a formula completely devoid of branched chain amino acids (contact metabolic dietitian) should be initiated as soon as possible by NG tube, to further prevent catabolism.

Please contact us immediately by calling 801 662-1000 and asking for the geneticist on call to be paged.

Thank you in advance.

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